

iPSCORE: A Resource of 222 iPSC Lines Enabling Functional Characterization of Genetic Variation across a Variety of Cell Types.

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Public Summary:

Large-scale collections of induced pluripotent stem cells (iPSCs) could serve as powerful model systems for examining how genetic variation affects biology and disease. Here we describe the iPSCORE resource: a collection of systematically derived and characterized iPSC lines from 222 ethnically diverse individuals that allows for both familial and association-based genetic studies. The iPSCORE collection contains representative individuals for risk and non-risk alleles for 95% of SNPs associated with human phenotypes through genome-wide association studies. Our study demonstrates the utility of iPSCORE for examining how genetic variants influence molecular and physiological traits in iPSCs and derived cell lines.

Scientific Abstract:

Large-scale collections of induced pluripotent stem cells (iPSCs) could serve as powerful model systems for examining how genetic variation affects biology and disease. Here we describe the iPSCORE resource: a collection of systematically derived and characterized iPSC lines from 222 ethnically diverse individuals that allows for both familial and association-based genetic studies. iPSCORE lines are pluripotent with high genomic integrity (no or low numbers of somatic copy-number variants) as determined using high-throughput RNA-sequencing and genotyping arrays, respectively. Using iPSCs from a family of individuals, we show that iPSC-derived cardiomyocytes demonstrate gene expression patterns that cluster by genetic background, and can be used to examine variants associated with physiological and disease phenotypes. The iPSCORE collection contains representative individuals for risk and non-risk alleles for 95% of SNPs associated with human phenotypes through genome-wide association studies. Our study demonstrates the utility of iPSCORE for examining how genetic variants influence molecular and physiological traits in iPSCs and derived cell lines.

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